

Erkennen von angeborenen Lipidstoffwechselstörungen

Prof. Martin Hersberger, Universitäts-Kinderspital Zürich, Abteilung für Klinische Chemie und Biochemie, Zürich

Literatur

- Handbuch der Fettstoffwechselstörungen, Schattauer Verlag, Stuttgart, 2007, 3. Auflage
- Family history and cardiovascular risk in familial hypercholesterolemia: data in more than 1000 children. Wiegman A, Rodenburg J, de Jongh S, Defesche JC, Bakker HD, Kastelein JJ, Sijbrands EJ. *Circulation* (2003) 107; 1473-1478
- Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. Wiegman A, Gidding SS, Watts GF, Chapman MJ, Ginsberg HN, Cuchel M, Ose L, Averna M, Boileau C, Borén J, Bruckert E, Catapano AL, Defesche JC, Descamps OS, Hegele RA, Hovingh GK, Humphries SE, Kovanen PT, Kuivenhoven JA, Masana L, Nordestgaard BG, Pajukanta P, Parhofer KG, Raal FJ, Ray KK, Santos RD, Stalenhoef AF, Steinhagen-Thiessen E, Stroes ES, Taskinen MR, Tybjærg-Hansen A, Wiklund O; European Atherosclerosis Society Consensus Panel. *European Heart Journal* doi:10.1093/eurheartj/ehv157
- Sitosterolemia: a gateway to new knowledge about cholesterol metabolism. Berge KE. *Ann Med.* 2003;35(7):502-11
- Genetic basis of sitosterolemia. Lee MH, Lu K, Patel SB. *Curr Opin Lipidol.* 2001 Apr;12(2):141-9
- Mutations in the human ATP-binding cassette transporters ABCG5 and ABCG8 in sitosterolemia. Heimerl S, Langmann T, Moehle C, Mauerer R, Dean M, Beil FU, von Bergmann K, Schmitz G. *Hum Mutat.* 2002 Aug;20(2):151
- Marked HDL deficiency and premature coronary heart disease. Schaefer EJ1, Santos RD, Asztalos BF. *Current Opinion in Lipidology* 2010, 21:289–297
- Differential diagnosis of familial high density lipoprotein deficiency syndromes. von Eckardstein A. *Atherosclerosis.* 2006 Jun;186(2):231-9

- Guidelines for the diagnosis and management of chylomicron retention disease based on a review of the literature and the experience of two centers. Noel Peretti, Agnès Sassolas, Claude C Roy, Colette Deslandres, Mathilde Charcosset, Justine Castagnetti, Laurence Pugnet-Chardon, Philippe Moulin, Sylvie Labarge, Lise Bouthillier, Alain Lachaux, Emile Levy. Orphanet Journal of Rare Diseases 2010, 5:24